



X-Plain™

Cystic Fibrosis

Reference Summary

Cystic fibrosis is a disease of the body's mucus glands that runs in families. About 30,000 Americans have cystic fibrosis at any given time. 12 million more Americans are carriers of cystic fibrosis, but are not affected by it.

Cystic fibrosis is a serious disease that affects the respiratory and digestive systems in children and young adults. The sweat glands and the reproductive system are also affected.



This summary is for people with cystic fibrosis and those carrying the disease who are not affected by it. Family and friends of cystic fibrosis

patients may find answers to questions also.

This summary will help you understand what cystic fibrosis is and what causes it. The symptoms, diagnosis, and treatment of cystic fibrosis are also explained.

Cystic Fibrosis

Cystic fibrosis, or CF, is an inherited disease of the body's mucus and sweat glands. CF does not go away and it gradually gets worse over time. CF is usually fatal. On average, people with cystic fibrosis live about 30 years.

Our bodies depend on special cells that secrete mucus and sweat in order to function normally. Our lungs have these special cells in them. The mucus the special cells secrete is watery and helps trap dirt and expel it to the outside.

Mucus also lubricates the respiratory, digestive, and reproductive systems, and prevents tissue from drying out or getting infected. Sweat is also secreted to the

outside of the body to keep us cool.

Mucus in patients with cystic fibrosis is very thick and collects in the intestines and lungs. The result is malnutrition, poor growth, numerous respiratory infections, breathing difficulties, and eventually, permanent lung damage. Lung disease is usually the cause of death in most patients.

People with cystic fibrosis lose large amounts of salt when they sweat. This can upset the balance of minerals in the blood, which may cause abnormal heart rhythms. Going into shock is a risk.

Causes

Diseases like cystic fibrosis that are inherited are called *genetic diseases*. *Genetic* comes from the word gene. Genes control traits, or functions, of the body. For instance, a gene controls how much insulin is in the body. Insulin controls the level of sugar in the body. Other genes control the color of the eyes.

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In cystic fibrosis, the patient has a defective CF gene. A defective CF gene makes it difficult for chloride to move through cells. This causes abnormal mucus secretions and salt loss in the body.

A CF gene is defective if a child inherits 2 abnormal genes, 1 from each parent. That is why even if parents do not have cystic fibrosis, their child could still have it. The parents are called cystic fibrosis carriers.

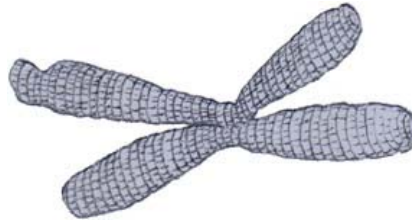
The human body is made up of millions of cells. Each cell has a nucleus.

Inside the cell nucleus are hereditary materials. The hereditary materials determine the traits and functions of the body. Each cell has identical hereditary materials because they all originate from the first cell produced when the sperm of the father fertilizes the egg of the mother.

There are 2 sets of hereditary materials: one from the mother and one from the father. When we look closely at the hereditary materials, they are made of chromosomes. Humans have 23 pairs of chromosomes: 1 set from the mother and 1 set from the father.

Chromosomes are actually instructions that tell the body what proteins to produce. Proteins make our body grow and function.

In chromosomes, series of instructions are clustered in areas called genes. A chromosome has thousands of genes.



Since we have 2 sets of chromosomes, we have a pair of genes for each trait. One from the father and one from the mother.

One gene from each pair is dominant over the other. For instance, if a child gets a black eye gene from her mother and a blue eye gene from her father and she has black eyes, then the black eye gene is dominant.

In cystic fibrosis, 2 genes control chlorine movement. One can be called regular CF gene G, and the other mutated CF gene g. Mutated means the gene was altered and became abnormal. Genes are sometimes altered accidentally in nature.

For the CF gene, the regular gene (G) is dominant over the mutated one (g). If a person has a pair of regular genes (GG), he or she does not carry the CF disease.

If a person has a pair including one regular and one mutated gene (Gg), he or she is a carrier of cystic fibrosis.

If a person has a pair of mutated genes (gg), he or she has cystic fibrosis. If a father and mother are both carriers of CF, what are the odds that their kids

- will NOT have cystic fibrosis?
- will be carriers of CF?
- will have cystic fibrosis?

Remember that each parent gives only 1 of their 2 CF genes during fertilization. Since both parents have Gg, each one can either give G or g.

If the father gives G and the mother gives G, the child is GG and does not have CF and does not carry it.

If the father gives g and the mother gives G, the child is gG, a CF carrier.

If the father gives G and the mother gives g, the child is Gg and a CF carrier.

If the father gives g and the mother gives g, the child is gg and has cystic fibrosis. For parents who are both carriers (Gg) there is a one in four chance that their baby will have cystic fibrosis.

Remember, when you toss a coin 4 times, you could get heads 4 times in a row! As a result, out of 4 children, parents who are carriers could have all children with CF, or all children without CF and not carriers!

Symptoms

Symptoms of cystic fibrosis vary from child to child. A baby born with cystic fibrosis usually has symptoms during its first year. However, sometimes signs do not show up until teen years or even later.

Infants or young children may have persistent diarrhea or bulky, foul-smelling, and greasy stools. Children may also have:

- frequent wheezing in the lungs
- pneumonia, a chronic cough with thick mucus
- poor growth

When babies are born with an intestinal blockage called *meconium ileus* there is a chance of cystic fibrosis. Since mucus in cystic fibrosis patients is very thick, it builds up quickly in the intestines and lungs. Symptoms of

cystic fibrosis are mostly due to thick mucus.

Cystic fibrosis can cause various other medical problems. These include

- inflamed sinuses, also called sinusitis
- nasal polyps, growths inside the nose
- rounded and enlarged fingers and toes
- coughing up blood

Other medical conditions associated with cystic fibrosis include

- rectal prolapse, when the rectum protrudes through the anus
- enlarged right side of the heart
- abdominal pain and discomfort
- too much gas in the intestine
- liver disease, diabetes, inflammation of the pancreas, and gallstones also occur in some patients with cystic fibrosis.

Diagnosis

After obtaining a detailed medical history and performing a thorough physical examination, the doctor may order some tests to try to diagnose cystic fibrosis with certainty.

The most common test for cystic fibrosis is called the sweat test. It measures the

amount of salt in the sweat. Patients with CF have a high amount of salt in their sweat. In the sweat test, an area of the skin, usually the forearm, is made to sweat by using a chemical and applying a mild electric current. To collect the sweat, the area is covered with a gauze pad or filter paper and wrapped in plastic.

After 30 - 40 minutes, the plastic is removed and the sweat on the pad or paper is analyzed. Higher than normal amounts of sodium and chloride suggest that the person may have cystic fibrosis.

The sweat test may not work well for newborns because they do not produce enough sweat. In that case, another type of test, called the immunoreactive trypsinogen test (IRT), may be used. In the IRT test, blood drawn 2 to 3 days after birth is analyzed for a specific protein called trypsinogen. Positive IRT tests must be confirmed by sweat and other tests.

A small percentage of people with cystic fibrosis have normal sweat chloride levels. They can only be diagnosed by chemical tests for the presence of the mutated gene.

Some other tests that can assist in the diagnosis of cystic fibrosis include

- chest x-rays
- lung function tests
- sputum (phlegm) cultures

Stool examinations can help identify the digestive abnormalities that are typical of cystic fibrosis.

Treatment

At present, there is no cure for cystic fibrosis. Treatment consists of reducing symptoms and slowing down the progress of the disease so the patient's quality of life is improved.

A major focus of cystic fibrosis treatment is to reduce breathing obstructions that cause frequent lung infections. Physical therapy, exercise, and medications are used to reduce the mucus blocking the airways.

For very advanced cystic fibrosis, lung transplant surgery may be an option. The digestive problems in cystic fibrosis are less serious and more easily managed than those in the lungs. A well-balanced, high-calorie diet, high in protein, and pancreatic enzymes that help digestion is often prescribed.

Supplements of vitamins A, D, E, and K are given to

ensure good nutrition. Enemas and medications that thin the mucus are used to treat intestinal obstructions.

Prevention

Currently, preventing cystic fibrosis is not possible once a baby is born. In babies with 2 abnormal CF genes, the disease is already present at birth.

Couples who have one or more children with CF in their families can have tests to determine if they are CF carriers. If they are, they are counseled regarding their odds of having another baby with CF.

A pregnant woman can have tests to find out if the baby she carries has cystic fibrosis. However, tests cannot detect all types of cystic fibrosis caused by different changes in the CF gene.

If there is another child with cystic fibrosis in the family, the expectant mother may request a prenatal test to see if the fetus has cystic fibrosis genes from both parents, is a carrier for one gene, or is altogether free of the cystic fibrosis genes.

There are 2 prenatal tests that can be done to check for CF: either an amniocentesis or chorionic villus biopsy. In

amniocentesis, cells from the amniotic fluid that surrounds the baby in the mother's womb are tested to see if CF genes similar to the parents' are present.

In chorionic villus biopsy, cells from the tissue that will eventually form the placenta are tested for the cystic fibrosis gene.

Facts

Learning about cystic fibrosis is critical for patients and their families in order to face the physical and emotional effects of the disease. Patients with CF can lead active lives. Parents should treat their children with cystic fibrosis as normally as possible. They should not be over-protective, but should encourage them to be active and self-reliant.

Cystic fibrosis is NOT contagious. People with CF can have a normal sex life. However, most, if not all, men are infertile.

Women with cystic fibrosis can have children, although they may be less fertile than women without cystic fibrosis.

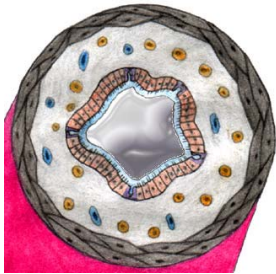
Although it affects all races and ethnic groups, cystic fibrosis occurs mostly in white people of northern European descent. It is less common in African-

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Americans, Native Americans and Asian Americans.

Summary

Cystic fibrosis is a chronic disease that mainly affects the lungs and the digestive system. The disease progresses and can be fatal. Thanks to significant advances in medicine, people with cystic fibrosis can live longer with a better quality of life. Scientists continue to learn more about the causes of cystic fibrosis and how to develop new treatments.



Patients and families should work closely with doctors and other health care professionals to develop self-management skills that can improve their quality of life.

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